Progressive anterior vertebral fusion: A report of three cases

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Abstract We report a series of three cases of progressive anterior vertebral fusion diagnosed and monitored in our establishment. This very rare condition was discovered in young children while exploring a spinal deformity. With X-rays of the thoracolumbar spine it was possible to make a positive diagnosis and ensure follow-up. The radiographic semeiotics are characteristic and combine anterior pinching of the disc, well-delimited erosion of the anterior vertebral corners and anterior then posterior intervertebral ankylosis. MRI is very useful for assessing the extent of the intervertebral ankylosis and the condition of the residual discs, without irradiation. The condition develops over several months or years. Conservative treatment is usually sufficient.

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Clinical cases

Observation 1

Louis had been monitored in paediatrics since the age of three months for congenital torticollis and gastro-oesophageal reflux. When he was one year old, his mother noticed the presence of thoracolumbar curvature. Clinical examination confirmed this curvature and the onset of thoracic lordosis overlying it. Physiological and biochemical tests were normal. X-ray of the spine revealed predominantly anterior pinching of the disc in many segments from T11 to L5, with incipient intervertebral ankylosis in certain segments and the onset of thoracolumbar kyphosis, encouraging PAVF (Fig. 1a). A follow-up X-ray performed 6 months later showed progression of the intervertebral ankylosis and of the kyphosis (Fig. 1a). It was decided that he should be
monitored clinically and radiologically. A CT scan performed a year later showed the extent of the ankylosis more precisely (Fig. 1b). There was also well-delimited erosion of the anterior part of the vertebral endplates and reactive osteosclerosis of the vertebral bodies. Two years later, an X-ray performed on EOS® (Biospace Imaging, Paris, France) showed relative stability of the bone lesions and that the kyphosis had not progressed (Fig. 1c). In MRI, the intervertebral discs appeared more or less mobile in the ankylosed segment. There was no associated intracanal abnormality. Since the clinical examination was normal and the kyphosis was not evolving, the recommendation was to simply monitor the situation.

**Figure 1.** Observation 1. a: X-ray of the thoracolumbar spine. Multi-segment anterior pinching of the disc with reactive osteosclerosis, incipient anterior intervertebral ankylosis and discrete kyphosis on the initial X-ray (M0). Six months later (M6), ankylosis has progressed and kyphosis is more pronounced; b: CT scan of the thoracolumbar spine (sagittal reconstructions in bone window and VRT mode). Bone changes in the vertebrae are clearly visible in the CT scan and consist of finely delimited erosion of the vertebral endplates, reactive osteosclerosis of the trabeculae and intervertebral ankylosis. Note the presence of intradiscal gas (arrow) indicating involvement of the disc. Anterior vertebral fusion is clearly visible on the VRT reconstruction; c: follow-up X-ray after 2 years; d: T2-weighted MRI of the thoracolumbar spine. In the lowest lumbar segment, only the posterior part of the intervertebral discs still has a normal signal. The higher discs have degenerated, in T2 hyposignal, and have partly disappeared because of ankylosis.

**Observation 2**

Lily had been monitored by paediatrics since her birth for right pulmonary hypoplasia due to scimitar syndrome. When she was four years old, a spinal deformity was discovered. Spinal X-ray images confirmed the presence of right convex thoracolumbar scoliosis, with no significant rotation of the vertebrae. There were also bone changes (erosion of the anterior vertebral corners, reactive osteosclerosis) extending from L1-L2 to L3-L4 reminiscent of PAVF. She was prescribed simple physiotherapy for the spine and thorax. At the age of nine, an imaging examination including X-rays and MRI (Fig. 2) showed anterior lumbar vertebral fusion.
we, on the MRI, signs of multi-segment disc alteration (T2 hyposignal). Clinical monitoring was decided.

Observation 3

Thomas had been monitored in paediatrics since he was seven for a right thoracic scoliosis. At the time of a follow-up X-ray check-up, incipient anterior intervertebral ankylosis was discovered by chance in L3-L4 (Fig. 3). MRI confirmed the onset of PAVF and objectified signal abnormalities from the anterior vertebral corners (Fig. 3). Simple physiotherapy was then prescribed as well as clinical monitoring.

Discussion

PAVF was first described in 1949 by Knutsson [1]. Since then, about 80 observations have been collated in the literature [1–6]. Since one series of 26 patients was reported in 1991 by the university hospital of Copenhagen [5], the term “Copenhagen syndrome” is also sometimes used to denote this condition. As our three observations show, PAVF is characterised by the occurrence in the child of evolving vertebral fusion in the thoracic and/or lumbar regions. One or more segments may be affected, whether adjacent or not [4]. The condition may be discovered by chance on X-rays, or when a spinal deformity (kyphosis, scoliosis) is noted, as in the three cases described earlier, or when there is often moderate spinal pain [4–6]. Medullary compression secondary to considerable kyphosis is possible [4], although exceptional. PAVF usually starts in the young child [4–6], affecting girls (60% of cases) slightly more than boys [5]. Its aetiology is still unknown. No infection has been found, which is why in English and American literature it is regularly called “progressive non-infectious anterior vertebral fusion” [3]. Genetic [3] and iatrogenic (antenatal exposure to thalidomide) [5] hypotheses have been proposed but have not been able to be confirmed. Al Kaissi et al. [7] have reported observation of PAVF of the cervical and thoracic spine and lumbosacral junction, associated with medullary abnormalities (tethered spinal cord, diastematomyelia) and with situs inversus, but the cervical spine is usually spared and the PAVF, isolated [1–5]. In the two cases of PAVF that underwent surgery, the anatomopathological examination revealed the presence of dystrophic cartilage (chondrocyte disorganisation containing areas of necrosis) anterior to the vertebral bodies, in continuity with the cartilaginous end-plate [3].

X-ray examination shows that PAVF starts with anterior pinching of the disc, associated with irregularities, well-delimited erosion (observation n°3) and reactive osteosclerosis of the vertebral corners. The posterior part of the intervertebral disc is relatively unaffected, at least at onset of the condition. The posterior arch is usually intact but not always [6]. In a few months (observation n°1) or years (observation n°2), ankylosis of the anterior part of the disc appears, then of its posterior part. Intervertebral ankylosis and the fact that the posterior arch is not affected then seem to be responsible for the appearance of kyphosis [4]. CT scans are of limited use, added to which the examination involves irradiation, but this technique does identify the specific vertebral bone changes (observation n°1). MRI is the examination of choice, firstly so that the precise extent of the intervertebral ankylosis can be seen and secondly so that the condition of the discs can be assessed [6], as illustrated by our three observations. T2-weighted sequences are in particular very useful for detecting disc alterations early on.
The associated abnormalities of the bone marrow of the vertebral endplates predominate at the vertebral corners and vary depending on the stage of the condition (oedema at the initial stage, fatty conversion at a later stage) [6]. Treatment of PAVF is usually conservative.

Imaging PAVF may bring other diagnoses into discussion such as infectious spondylodiscitis, congenital vertebral block and Scheuermann’s disease. Infectious spondylodiscitis arises in a very different clinical context (alteration in the general condition, fever, pain, spinal stiffness, etc.). Physiological and biochemical tests indicate an infection. It is possible for several segments to be affected but this is rarer than involvement of just one segment. Radiography shows erosion of the vertebral endplates, which is indistinct, and poorly delineated. MRI objectifies inflammation of the bone marrow of the vertebral endplates and in particular, infiltration of the soft tissues and/or infectious epiduritis. Evolution towards vertebral fusion is possible but does not occur as a rule. Congenital vertebral fusion is a defect of spinal segmentation. It can involve one or, more rarely, several segments, whether adjacent or not. It concerns the vertebral body and sometimes, the posterior arch, unlike PAVF. There may be costal synostosis in the thoracic region. This type of vertebral fusion is present from birth and is accompanied by anterior vertebral concavity on the X-rays, indicating that it is congenital. In addition, on successive X-rays, the vertebral fusion remains unchanged. The diagnoses of infectious spondylodiscitis and congenital vertebral fusion could easily be ruled out in the three observations reported above. Scheuermann’s disease (or vertebral epiphysitis) was the principle differential diagnosis. This is juvenile spinal osteochondrosis occurring on a cartilaginous vertebral endplate made fragile by repeated microtrauma (the demands of sport, obesity) [8]. It arises at puberty [8,9]. The mid and lower thoracic spine is classically affected (at least three contiguous vertebrae involved) and sometimes, the lumbar spine. Exclusively lumbar involvement is rare. It is considered to be a particular form of Scheuermann’s disease (lumbar Scheuermann’s disease), or a distinct entity (juvenile lumbar osteochondrosis), and is particularly encountered in adolescents practising high performance sport [9,10]. As in PAVF, X-rays of the spine reveal irregularities of the vertebral endplates and predominantly anterior pinching of the intervertebral discs. Ossification of the anterior part of the intervertebral discs may also result in anterior vertebral fusion [9,10] but other X-ray anomalies are specific to Scheuermann’s disease: wedging of the vertebral bodies with anterior translation (at least 5°), upper thoracic kyphosis of 40°, peripheral or central intraspinal herniation, increase in the anteroposterior diameter and reduction in height of the vertebral bodies [9–11]. In practice, the age of the child (usually under 10), the clinical context (few or no symptoms), the imaging appearance (anterior pinching of the disc, well-defined erosion, reactive osteosclerosis, anterior intervertebral ankylosis), and the fact that the lesions are evolving are just so many arguments in favour of PAVF, as highlighted by our three observations.

Conclusion

PAVF characteristically occurs in children and involves progressive intervertebral ankylosis. The diagnosis can be established from X-rays of the thoracolumbar spine, which also assist in monitoring the patient. MRI is very useful for analysing the extent of the intervertebral ankylosis and assessing the condition of the discs. Although a very rare condition, we should be aware of PAVF for there to be no problem diagnosing it from the imaging and so that conservative treatment can be initiated in the guise of clinical monitoring during bone growth.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

References